# WEB-BASED RESOURCES IN OPHTHALMOLOGY:
## RESOURCES & STRATEGIES

Pamela C. Sieving, MA, MS
National Institutes of Health
Bethesda, Maryland

EVER 2006

# OBJECTIVES

- Medline and PubMed
- Other bibliographic databases
- Google Scholar, Windows Live
- Citations and Impact Factors
- Evidence-based medicine resources
- Genetics & bioinformatics for you (and your patients)
- Institutional Repositories

“An old tradition and a new technology have converged to make possible an unprecedented public good”

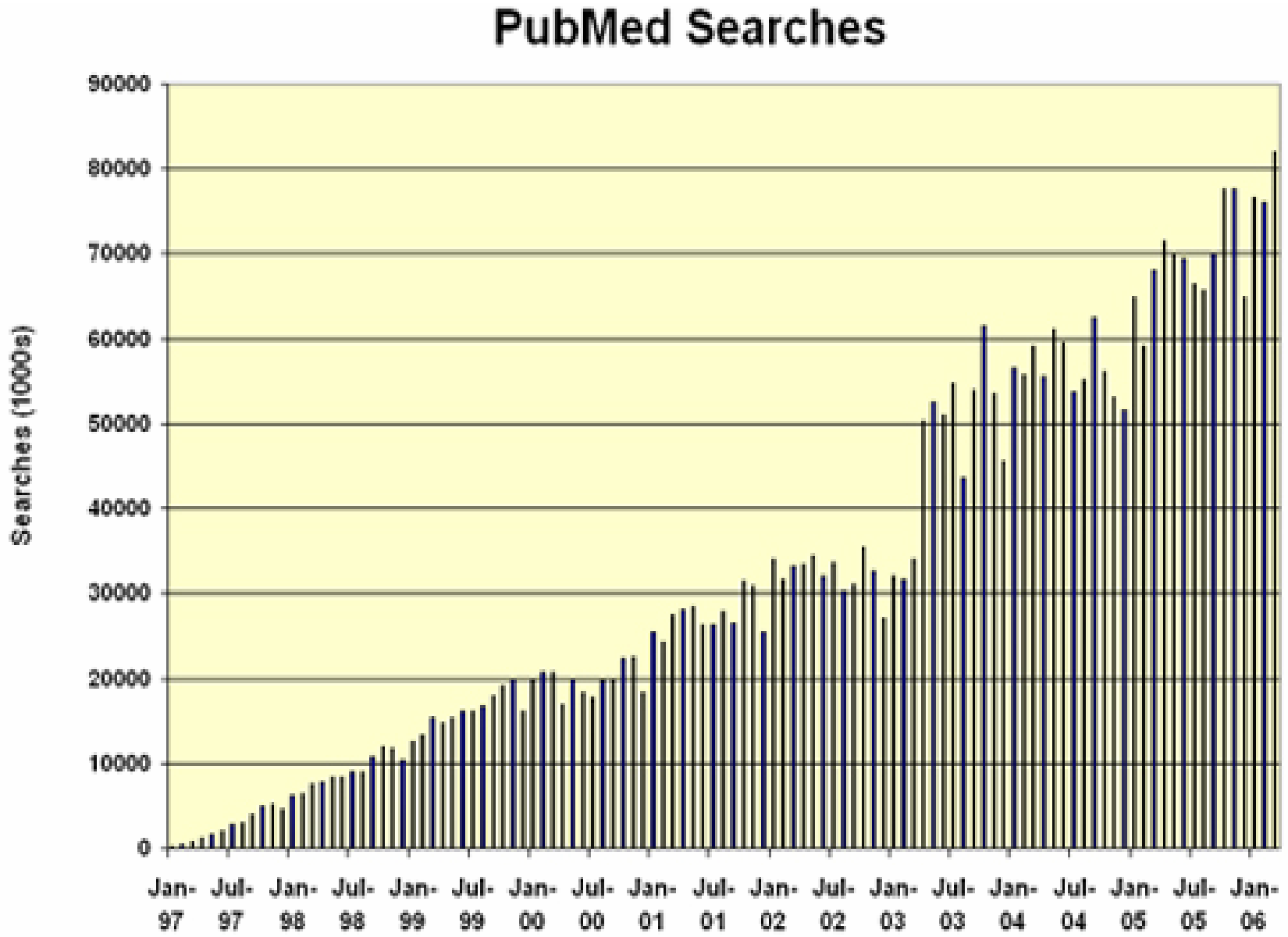
Budapest Open Access Initiative, 2002

# MEDLINE

- Index Medicus electronic, 1966—

# PubMed

- Medline
- In-process Medline records
- “Old Medline”
- Additional content
- Links
  - Genetics files
  - Books
- Related Records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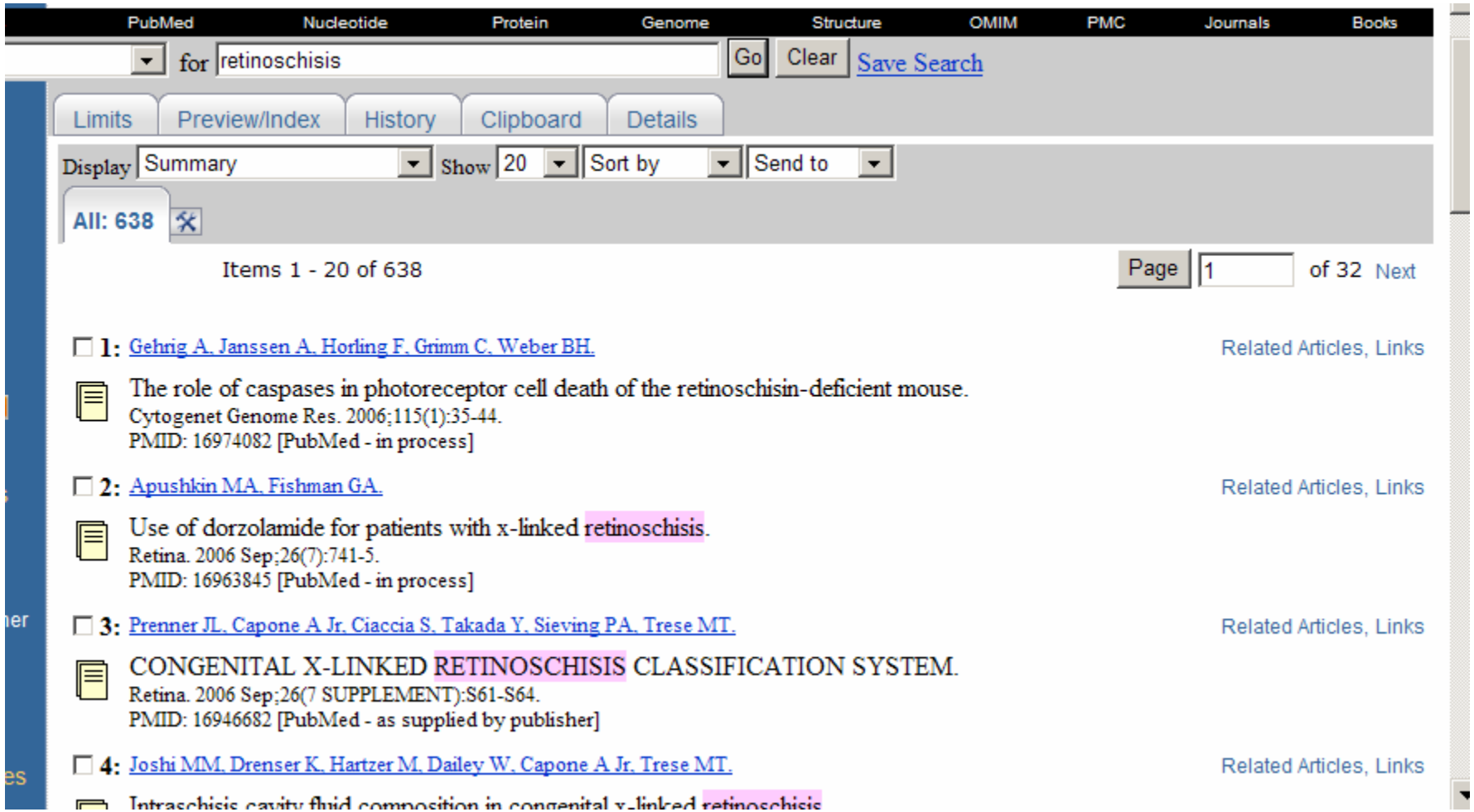
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Page 1 of 32 Next
1: Gehrig A, Janssen A, Horling F, Grimm C, Weber BH.
Related Articles, Links
The role of caspases in photoreceptor cell death of the retinoschisin-deficient mouse.
Cytogenet Genome Res. 2006;115(1):35-44.
PMID: 16974082 [PubMed - in process]
2: Apushkin MA, Fishman GA.
Related Articles, Links
Use of dorzolamide for patients with x-linked retinoschisis.
Retina. 2006 Sep;26(7):741-5.
PMID: 16963845 [PubMed - in process]
3: Prenner JL, Capone A Jr, Ciaccia S, Takada Y, Sieving PA, Trese MT.
Related Articles, Links
CONGENITAL X-LINKED RETINOSCHISIS CLASSIFICATION SYSTEM.
Retina. 2006 Sep;26(7 SUPPLEMENT):S61-S64.
PMID: 16946682 [PubMed - as supplied by publisher]
4: Joshi MM, Drenser K, Hartzer M, Dailey W, Capone A Jr, Trese MT.
Related Articles, Links
Intraschisis cavity fluid composition in congenital x-linked retinoschisis

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1: Cytogenet Genome Res. 2006;115(1):35-44.

KARGER Full Text | Links

**The role of caspases in photoreceptor cell death of the retinoschisin-deficient mouse.**

**Gehrig A**, **Janssen A**, **Horling F**, **Grimm C**, **Weber BH**.

Institute of Human Genetics, University of Regensburg, Regensburg, Germany.

Early schisis cavities in the retinal bipolar cell layer accompanied by progressive loss of cone and rod photoreceptor cells are the hallmark of the retinoschisin-deficient (Rs1h(-/Y)) murine retina. With this study we aimed at elucidating the molecular events underlying the photoreceptor cell death in this established murine model of X-linked juvenile retinoschisis. We show that photoreceptor degeneration in the Rs1h(-/Y) mouse is due to apoptotic events peaking around postnatal day 18. Cell death is accompanied by increased expression of initiator and inflammatory caspases but not by downstream effector caspases. The strong induction of caspase-1 (Casp1) prompted us to explore its involvement in the apoptotic process. We therefore generated double knock-out mice deficient for both retinoschisin and Casp1. No direct influence of the Casp1 genotype on apoptosis could be identified although striking differences in the overall number of resident microglia were observed independent of the Rs1h genotype. 

PMID: 16974082 [PubMed - in process]

Related Links

- RS-1 Gene Delivery to an Adult Rs1h Knockout Mouse Model Restores ERG b-W [Invest Ophthalmol Vis Sci. 2004]
- Caspase-3 inhibitor reduces apototic photoreceptor cell death during inherited retinal degen [Mol Vis. 2003]
- Inactivation of the murine X-linked juvenile retinoschisis gene, Rs1h, [Proc Natl Acad Sci U S A. 2002]
- Multiple, parallel cellular suicide mechanisms participate in photoreceptor cell de: [Exp Eye Res. 2006]
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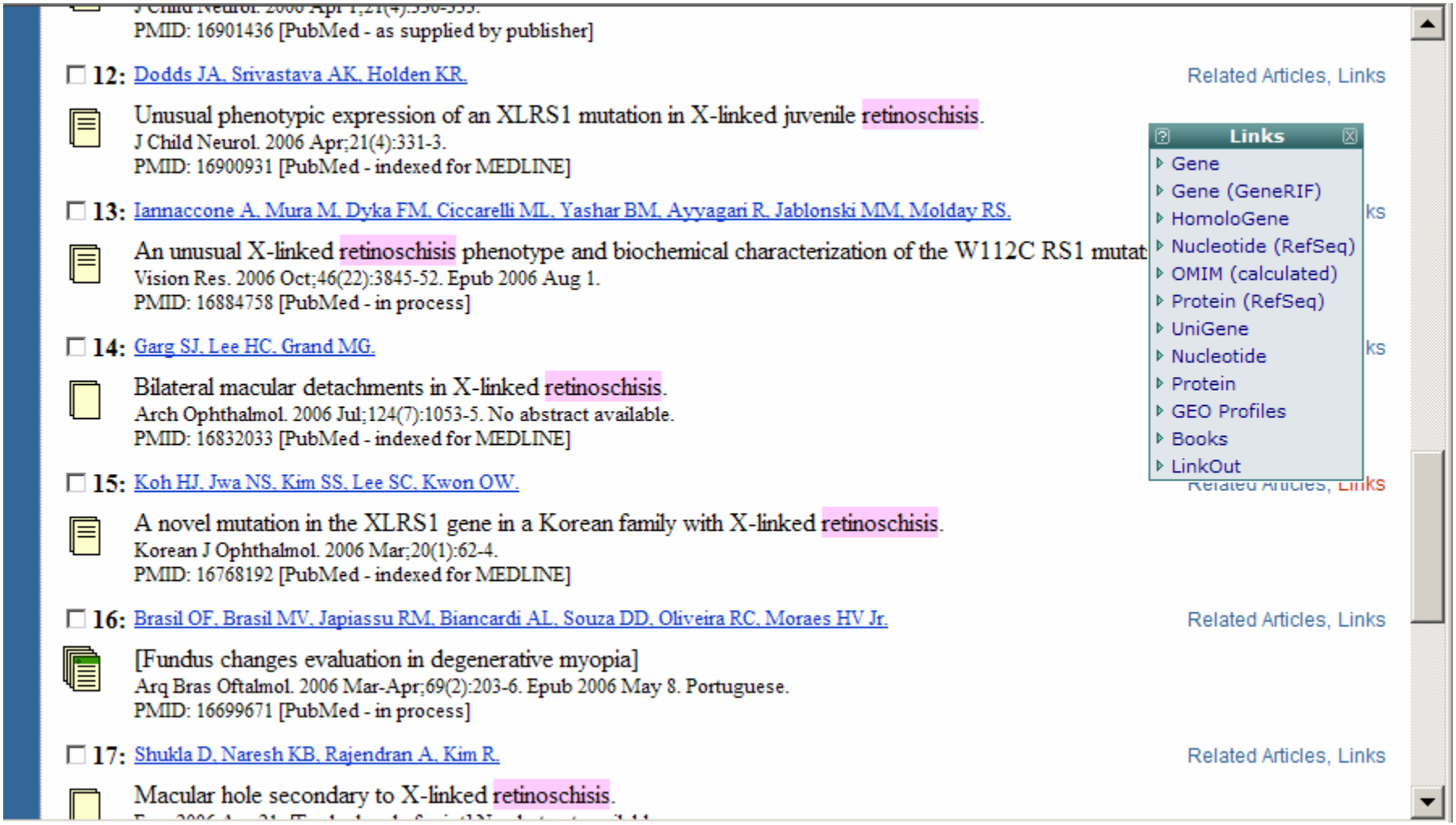
PMID: 16901436 [PubMed - as supplied by publisher]

12: Dodds JA, Srivastava AK, Holden KR. Related Articles, Links

Unusual phenotypic expression of an XLRS1 mutation in X-linked juvenile retinoschisis.
J Child Neurol. 2006 Apr;21(4):331-3.
PMID: 16900931 [PubMed - indexed for MEDLINE]

13: Iannaccone A, Mura M, Dyka FM, Ciccarelli ML, Yashar BM, Ayyagari R, Jablonski MM, Molday RS. ks

An unusual X-linked retinoschisis phenotype and biochemical characterization of the W112C RS1 mutat
Vision Res. 2006 Oct;46(22):3845-52. Epub 2006 Aug 1.
PMID: 16884758 [PubMed - in process]

14: Garg SJ, Lee HC, Grand MG. ks

Bilateral macular detachments in X-linked retinoschisis.
Arch Ophthalmol. 2006 Jul;124(7):1053-5. No abstract available.
PMID: 16832033 [PubMed - indexed for MEDLINE]

15: Koh HJ, Jwa NS, Kim SS, Lee SC, Kwon OW. Related Articles, Links

A novel mutation in the XLRS1 gene in a Korean family with X-linked retinoschisis.
Korean J Ophthalmol. 2006 Mar;20(1):62-4.
PMID: 16768192 [PubMed - indexed for MEDLINE]

16: Brasil OF, Brasil MV, Japiassu RM, Biancardi AL, Souza DD, Oliveira RC, Moraes HV Jr. Related Articles, Links

[Fundus changes evaluation in degenerative myopia]
Arq Bras Oftalmol. 2006 Mar-Apr;69(2):203-6. Epub 2006 May 8. Portuguese.
PMID: 16699671 [PubMed - in process]

17: Shukla D, Naresh KB, Rajendran A, Kim R. Related Articles, Links

Macular hole secondary to X-linked retinoschisis.

Links
- Gene
- Gene (GeneRIF)
- HomoloGene
- Nucleotide (RefSeq)
- OMIM (calculated)
- Protein (RefSeq)
- UniGene
- Nucleotide
- Protein
- GEO Profiles
- Books
- LinkOut

☐ **39:** Ophthalmic Genet. 2005 Sep;26(3):111-7. Related Articles, Links

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**Juvenile X-linked retinoschisis with normal scotopic b-wave in the electroretinogram at an early stage of the disease.**

**Eksandh L**, **Andreasson S**, **Abrahamson M**.

Department of Ophthalmology, University Hospital of Lund, Sweden. louise.eksandh@telia.com

PURPOSE: To report four cases of genetically verified juvenile X-linked retinoschisis (XLRS) with normal scotopic b-waves in full-field ERG, including one patient with a novel mutation (W50X) in the RS1 gene. METHODS: Four XLRS patients from different families were examined with regard to visual acuity, kinetic perimetry, fundus photography, full-field ERG, and OCT. Two of these patients were also examined with multifocal-ERG (mfERG). Mutations in the RS1 gene were identified by sequence analysis. RESULTS: The full-field ERG presented normal b-wave amplitudes on scotopic white-light stimulation. OCT and mfERG presented macular schisis and macular dysfunction. Genetic analysis revealed a deletion of exon 1 and the promotor region in one patient and mutations giving rise to the amino acid substitutions R209C and W96R in two others. The fourth patient carried a novel mutation in exon 3 of the RS1 gene (nt 149 G-->A), causing the introduction of a stop codon after amino acid 49 in the RS protein. CONCLUSION: Four young males with XLRS did not present with reduction in the scotopic b-wave amplitude on full-field ERG, which is otherwise often considered to be characteristic of the disease. Full-field ERG and molecular genetic analysis of the RS1 gene still remain the most important diagnostic tools for this retinal disorder, although the OCT can be a valuable complement in order to make the diagnosis at an early stage.

MeSH Terms:

gene still remain the most important diagnostic tools for this retinal disorder, although the OCT can be a valuable order to make the diagnosis at an early stage.

MeSH Terms:

- Child
- DNA Mutational Analysis
- Dark Adaptation
- Electroretinography*
- Exons/genetics
- Eye Proteins/genetics
- Humans
- Male
- Mutation
- Perimetry
- Photic Stimulation
- Photoreceptors, Vertebrate/physiology*
- Research Support, Non-U.S. Gov't
- Retinoschisis/genetics
- Retinoschisis/physiopathology*
- Tomography, Optical Coherence
- Visual Acuity

Substances:

- Eye Proteins
- RS1 protein, human

# genetics

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# OMIM Statistics for September 29, 2006

## Number of Entries

| | Autosomal | X-Linked | Y-Linked | Mitochondrial | **Total** |
|---|---|---|---|---|---|
| * Gene with known sequence | 10447 | 483 | 48 | 37 | 11015 |
| + Gene with known sequence and phenotype | 351 | 33 | 0 | 0 | 384 |
| # Phenotype description, molecular basis known | 1795 | 157 | 2 | 26 | 1980 |
| % Mendelian phenotype or locus, molecular basis unknown | 1415 | 137 | 4 | 0 | 1556 |
| Other, mainly phenotypes with suspected mendelian basis | 2029 | 144 | 2 | 0 | 2175 |
| **Total** | 16037 | 954 | 56 | 63 | **17110** |

# What is a digital resource?

- electronic books
- online journals
- audio files (e.g., wav, mp3)
- images (e.g., tiff, gif)
- movies (e.g., mpeg, quicktime)
- reference texts (e.g., dictionaries, directories)
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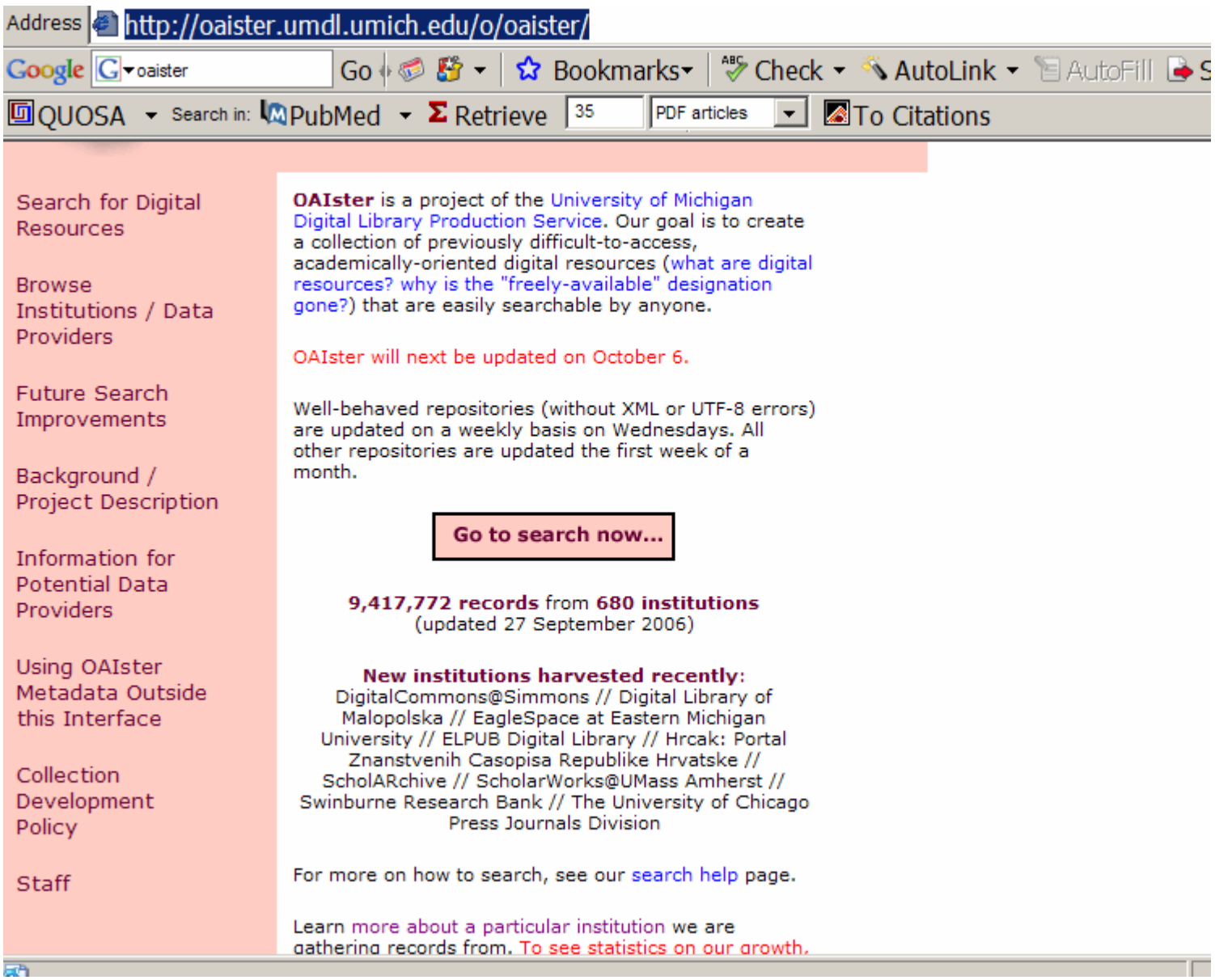
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OAIster is a project of the University of Michigan Digital Library Production Service. Our goal is to create a collection of previously difficult-to-access, academically-oriented digital resources (what are digital resources? why is the "freely-available" designation gone?) that are easily searchable by anyone.
OAIster will next be updated on October 6.
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| | |
|---|---|
| Title | Evaluation of the visual pathway with ERG, mfERG and mfVEP in inherited eye disorders |
| Author/Creator | Gränse, Lotta |
| Publisher | Lunds universitet/Dept of Ophthalmology, **Lund** University |
| Year | 2006-03-24 |
| Resource Type | text.thesis.doctoral |
| Language | English |
| Note | This thesis will describe the clinical phenotypes, with emphasis on electrophysiology, in patients with different hereditary eye diseases and to further evaluate and modify the mfVEP technique for clinical use. Bothnia Dystrophy is a tapetoretinal disorder with a mutation in the RLBP1 gene. Early in the disease the fundus may have a normal appearance. The full-field ERG demonstrates an absence of the rod response but normal amplitudes for the cones. However, after prolonged dark adaptation the rods recover completely. MfERG can be used for objective documentation of the disturbed **macular** function. Patients with retinitis pigmentosa may not always follow the typical natural course of the disorder with progressive loss of the central visual fields, which may in some patients remain unaffected for several decades. MfERG and mfVEP may be of clinical use in evaluating remaining visual function in these patients. Patients with dominant optic atrophy and a known mutation in the OPA-1 gene have a very variable clinical phenotype. MfVEP and ocular blood flow measurements are two new methods for improved identification and characterization of this disorder. A patient with a known mutation for Leber`s hereditary optic neuropathy (LHON) was followed during the acute stage of the disease with mfVEP, demonstrating a correlation to the progression of the disease. The mfVEP may be of clinical value as an objective method for monitoring the course of this disease. MfVEP demonstrates the cortical response corresponding to the central visual field. An improvement for the clinical value of the method was the use of an IR-camera for both stimulation and for controlling the fixation. By introducing a two channel system it was possible to describe the uncrossed/crossed visual pathways and analyze inter-ocular differences. |
| Subject | Oftalmologi; Full-field ERG; multifocal ERG; multifocal VEP; Bothnia Dystrophy; Retinitis pigmentosa; Dominant optic atrophy; Leber`s hereditary optic neuropathy |
| URL | http://theses.lub.lu.se/postgrad/search.tkl?field_query1=pubid&query1=med_1266&recordformat=display |
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| Subject | REPORTS |
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| Title | **Retinoschisis**: Perimetry as a Clue to Diagnosis |
| Author/Creator | Sullivan, Garrett L.; von Pirquet, Silvio R. |
| Resource Type | Text |
| Language | English |
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| URL | http://www.pubmedcentral.gov/articlerender.fcgi?artid=1316399 |
| Institution | PubMed Central (PMC) |

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| Author/Creator | 佐藤, 正樹 |
| Contributor | Sato, Masaki |
| Contributor | サトウ, マサキ |

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Trans Am Ophthalmol Soc

Trans Am Ophthalmol Soc. 1961; 59: 80–95.



# Retinoschisis: Perimetry as a Clue to Diagnosis

Garrett L. Sullivan and Silvio R. von Pirquet



## Full text

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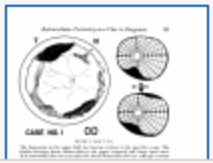

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